

AMENDMENTS TO THE CLAIMS

The listing of claims will replace all prior versions, and listings, of claims in the application. Please amend Claims 109-113, 115, 117-120 and 151-153 as follows:

Listing of Claims:

Claims 1-108 (canceled)

Claim 109 (currently amended): A nucleic acid probe or primer comprising[[ : ]]  
~~(A) a nucleotide sequence selected from the group consisting of:~~  
~~(A) (SEQ ID NO:1), a nucleotide sequence complementary thereto, a degenerate coding sequence thereof, or a gene specific fragment of any of these (SEQ ID NO:3), (SEQ ID NO:4), (SEQ ID NO:5), or (SEQ ID NO:6); or~~  
~~(B) a sequence fully complementary to any of (A);~~  
~~(C) a degenerate coding sequence of any of (SEQ ID NO:1), (SEQ ID NO:3) or (SEQ ID NO:5); and~~  
~~(D) a nucleic acid segment encoding a human PAPSS2 protein having an amino acid sequence of (SEQ ID NO:7).~~

Claim 110 (currently amended): The nucleic acid probe or primer of Claim 109, wherein the ~~gene specific fragment has a nucleotide sequence comprising~~ is 5'-  
TGGACCAAGGATGACGATGT-3' (SEQ ID NO:[ [ ]3]), a sequence fully complementary nucleotide sequence ~~thereto, or a PAPSS2 specific sequence overlapping either of these at 5 or more contiguous nucleotides at its 5' or 3' end~~ a degenerate coding sequence of (SEQ ID NO:3).

Claim 111 (currently amended): The nucleic acid probe or primer of Claim 109, wherein the ~~gene specific fragment has a nucleotide sequence comprising~~ is 5'-  
CGGAAAGATGGCAACAATGG (SEQ ID NO:[ [ ]4][ [, ]]) or a sequence fully complementary nucleotide sequence, or a PAPSS2 specific sequence overlapping either of these at 5 or more contiguous nucleotides at its 5' or 3' end ~~thereto.~~

Claim 112 (currently amended): The nucleic acid probe or primer of Claim 109, wherein the ~~the gene specific fragment has a nucleotide sequence comprising~~ is 5'-CTGGTGCTGGAAAAACAAACG-3' (SEQ ID NO:[ ]5), a sequence fully complementary nucleotide sequence ~~ethereto~~, or a ~~PAPSS2 specific sequence overlapping either of these at 5 or more contiguous nucleotides at its 5' or 3' end~~ a degenerate coding sequence of (SEQ ID NO:5).

Claim 113 (currently amended): The nucleic acid probe or primer of Claim 109, wherein the ~~the gene specific fragment has a nucleotide sequence comprising~~ is 5'-TGCAGATGGAGAAATAAAGCTG (SEQ ID NO:[ ]6)[,] or a sequence fully complementary nucleotide sequence, or a ~~PAPSS2 specific sequence overlapping either of these at 5 or more contiguous nucleotides at its 5' or 3' end~~ thereto.

Claim 114 (canceled)

Claim 115 (currently amended): An oligonucleotide primer for amplifying a PAPSS2-specific nucleic acid segment, comprising a nucleotide sequence selected from the group consisting of:

- (A) (SEQ ID NO:3), (~~SEQ ID NO:4~~), (SEQ ID NO:5), (SEQ ID NO:6), (SEQ ID NO:11), (SEQ ID NO:12), (SEQ ID NO:13), (SEQ ID NO:14), (SEQ ID NO:15), (SEQ ID NO:16), (SEQ ID NO:17), (SEQ ID NO:18), or (SEQ ID NO:28);
- (B) a nucleotide sequence fully complementary to any of (A); and
- (C) a PAPSS2-specific fragment of (A) or (B) at least 15 nucleotides long; or
- (D) a ~~PAPSS2 specific nucleotide sequence overlapping at 5 or more contiguous nucleotide positions any sequence of (A) or (B) at its 5' or 3' end.~~

Claim 116 (canceled)

Claim 117 (currently amended): A pair of oligonucleotide primers comprising a forward and a reverse primer, said pair capable of producing detectable nucleic acid amplification products having:

- (A) (SEQ ID NO:1) or (SEQ ID NO:9);
- (B) a nucleotide sequence fully complementary to any of (A); or
- (C) a PAPSS2 gene-specific fragment of (A) or (B).

Claim 118 (currently amended): The pair of oligonucleotide primers of Claim 117, wherein

the forward primer has a nucleotide sequence comprisingconsisting of 5'-TGGACCAAGGATGACGATGT-3' (SEQ ID NO:[ ]3), a fully complementary nucleotide sequence, or a PAPSS2-specific fragment of either of these at least 15 nucleotides long; and

the reverse primer has a nucleotide sequence comprisingconsisting of 5'-CGGAAAGATGGCAACAATGG-3' (SEQ ID NO:[ ]4), or a fully complementary nucleotide sequence, ~~or a PAPSS2-specific fragment of either of these at least 15 nucleotides long.~~

Claim 119 (currently amended): The pair of oligonucleotide primers of Claim 117, wherein

the forward primer has a nucleotide sequence comprisingconsisting of 5'-CTGGTGCTGGAAAAACAAACG-3' (SEQ ID NO:[ ]5), a fully complementary nucleotide sequence, or a PAPSS2-specific fragment of either of these at least 15 nucleotides long; and

the reverse primer has a nucleotide sequence comprisingconsisting of 5'-TGCAGATGGAGAAATA AAGCTG-3' (SEQ ID NO:[ ]6), a fully complementary nucleotide sequence, or a PAPSS2-specific fragment of either at least 15 nucleotides long.

Claim 120 (currently amended): The pair of oligonucleotide primers of Claim 117, wherein

the forward primer ~~comprises~~ has a nucleotide sequence consisting of:

- (A) (SEQ ID NO:3), (SEQ ID NO:5), (SEQ ID NO:11), (SEQ ID NO:12), or (SEQ ID NO:13);
- (B) a nucleotide sequence fully complementary to any of (A); and
- (C) a ~~gene-specific~~PAPSS2-specific fragment of (A) or (B) at least 15 nucleotides long;  
~~or~~  
(D) a ~~PAPSS2-specific~~ nucleotide sequence overlapping at 5 or more contiguous nucleotide positions ~~any sequence of (A) or (B)~~ at its 5' or 3' end; and  
[[a]] the reverse primer ~~comprising~~ has a nucleotide sequence consisting of:
  - (E)(D) (SEQ ID NO:4), (SEQ ID NO:6), (SEQ ID NO:14), (SEQ ID NO:15), (SEQ ID NO:16), (SEQ ID NO:17), or (SEQ ID NO:18);
  - (F)(E) a nucleotide sequence fully complementary to any of (E)(D); and
  - (G)(F) a PAPSS2-specific fragment of (E)(D) or (F)(E) at least 15 nucleotides long; or
  - (H) a ~~PAPSS2-specific~~ nucleotide sequence overlapping at 5 or more contiguous nucleotide positions ~~any sequence of (E) or (F)~~ at its 5' or 3' end.

Claims 121-150 (canceled)

Claim 151 (currently amended): A genetic testing kit for diagnosing spondyloepimetaphyseal dysplasia (SEMD) in a human subject or for identifying a human carrier of SEMD, said kit comprising an oligonucleotide primer(s) comprising a nucleotide sequence selected from the group consisting of:

- (A) a nucleotide sequence of (SEQ ID NO:3), (SEQ ID NO:4), (SEQ ID NO:5), (SEQ ID NO:6), (SEQ ID NO:[ ]11), (SEQ ID NO:[ ]12), (SEQ ID NO:13), (SEQ ID NO:14), (SEQ ID NO:15), (SEQ ID NO:16), (SEQ ID NO:17), (SEQ ID NO:18), or (SEQ ID NO:28);
- (B) a nucleotide sequence fully complementary to any of (A); and
- (C) a PAPSS2-specific fragment of (A) or (B) at least 15 nucleotides long; ~~or~~  
(D) a ~~PAPSS2-specific~~ nucleotide sequence at least 15 nucleotides long and overlapping at 5 or more contiguous nucleotide positions ~~any sequence of (A) or (B)~~ at its 5' or 3' end; and

instructions for using the primer(s) in diagnosing SEMD in a human subject or for identifying a human carrier of SEMD.

Claim 152 (currently amended): A genetic testing kit for diagnosing spondyloepimetaphyseal dysplasia (SEMD) in a human subject or for identifying a human carrier of ~~spondyloepimetaphyseal dysplasia~~SEMD, comprising the pair[[s]] of oligonucleotide primers of Claim 117; and

instructions for using the primer[[()s[()]]] in diagnosing SEMD in a human subject or for identifying a human carrier of SEMD.

Claim 153 (currently amended): A genetic testing kit for diagnosing spondyloepimetaphyseal dysplasia (SEMD) in a human subject, or for identifying a human carrier of ~~spondyloepimetaphyseal dysplasia~~SEMD, comprising the pair of oligonucleotide primers of Claim 120; and

instructions for using the primer[[()s[()]]] in diagnosing SEMD in a human subject or for identifying a human carrier of SEMD.

**AMENDMENTS TO DRAWINGS**

The attached sheets of drawings include formal renderings of Figures 1-4. The first sheet, which contains Figure 1, replaces the original sheet that included both Figures 1-2, and incorporates a formal rendering of the figure labeling to replace the handwritten label of Figure 1.

The second sheet contains Figure 2. In Figure 2, the previously omitted sequence designation, SEQ ID NO:29, has been inserted as well as a formal rendering of the figure labeling to replace the handwritten label of Figure 2. The sequence designation SEQ ID NO:29 was inserted into the brief description of Figure 2 in the *Response to Office Communication and Preliminary Amendment*, which Applicant mailed on October 24, 2002.

The third sheet is Figure 3, which replaces the original sheet of Figure 3. In Figure 3, the sequence designations, SEQ ID NOS:30-33, have been inserted as well as a formal rendering of the figure labeling to replace the handwritten label of Figure 3. Sequence designations SEQ ID NOS:30-33 were inserted into the brief description of Figure 3 in the *Response to Office Communication and Preliminary Amendment*, which Applicant mailed on October 24, 2002.

The fourth sheet is Figure 4, which now has a formal rendering of the figure labeling to replace the handwritten label of Figure 4.

No new matter was added by any of the above noted amendments to the figures.

Attachments (after page 17 and **Exhibit A**): Replacement Sheets 1-4